A Rare Case of Spondylo - Epiphyseal Dysplasia Congenita : Case Report

Nikhil Nagre[1], Vinod Khandait[2]

ABSTRACT

Spondylo - Epiphyseal Dysplasia Congenita (SED) is a rare form of skeletal systemic disease, characterized by congenital dwarfism with a short trunk and epiphysial dysplasia in the long bones and vertebral bodies. Patients also frequently suffer from atlanto-axial instability due to osodontoideum. Compression of the spinal cord caused by atlanto-axial instability is a common, serious complication in SED patients, and causes severe spinal cord symptoms or occasionally sudden death. We present an SED patient who presented with severe spinal cord symptoms due to atlanto-axial instability with incidental left gross HUN and starvation ketosis.

Introduction :

Spondylo-epiphyseal dysplasia congenita (SED) is a rare form of skeletal systemic disease, characterized by congenital dwarfism with a short trunk and epiphysial dysplasia in the long bones and vertebral bodies. In earlier studies, this disease was confused with Morquio's Disease (MPS type 4). In 1970, Spranger and Wiedemann established the disease concept with their report on the clinical and radiographic features of 29 cases of SED[1,2]. Since SED is frequently accompanied by atlantoaxial instability due to Osodontoideum[1], which causes severe spinal cord symptoms or occasionally sudden death, its prognosis and neurological recovery depend on early diagnosis and the timing of fusion and decompression procedures.

Case Presentation :

19 year old female patient residing from rural area in Gadchiroli district, presented with complaints of weakness of all 4 limbs, tingling sensation (paraesthesia) all over body below neck, 2 days prior to admission. Patient had history of similar weakness and paraesthesia that was transient and which improved spontaneously on two occasions, 5 days prior to admission. This time the patient's weakness was persistent and was associated with breathlessness and hence she was referred to Government Medical College, Nagpur for evaluation and management. Weakness had gradually progressed and patient was bed ridden on admission. Past history of progressive dyspnea on exertion was present since the past 1 year. There was history of renal calculi diagnosed 10 years back. She attended menarche at the age of 18 years along with appearance of secondary sexual characteristics. She was short in stature since childhood, Height 90 cm, Weight -15 kg, BP was 120/80 mmHg, Patient had dolichocephaly, depressed nasal bridge, webbed neck, barrel shaped chest, widened wrist with increased joint mobility. Neurological examination revealed hypoesthesia below C2, hyperactive deep reflexes in both upper and lower limbs, plantars flexor. Power 4+ in upper limb and 3+ in lower limb with normal intelligence.

Her RBS on admission was 52 mg% with large urine ketones - large on admission. She was treated as starvation ketosis. Later her fasting and postprandial sugars were normal with HBA1C of 4.6. USG abdomen + pelvis was done s/o left sided gross Hydro-Uretero-Nephrosis,CT KUB was done s/o gross left HUN with few obstructing calculi in middle1/3 and distal segment of left ureter. Urology opinion taken regarding the same and USG guided left PCN was done. 2D-Echo was normal.

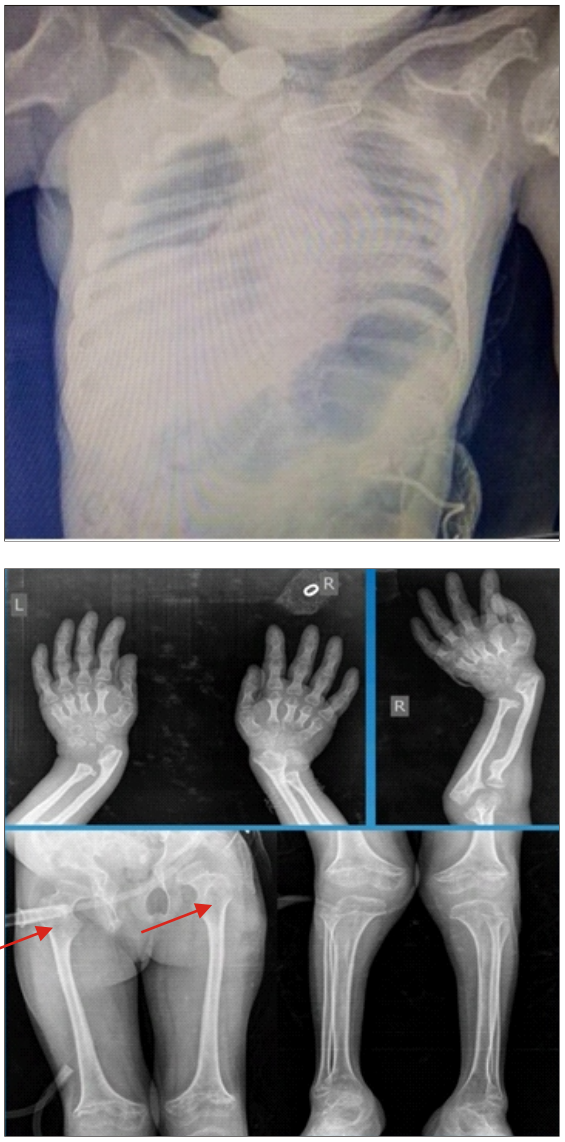
CT whole spine study s/o platyspondyly (flat vertebral bodies),exaggeration of cervical and lumbar lordosis, os odontoideum, atlanto-axial subluxation, flattening of femoral heads and greater trochanter.

[1]Resident, [2]Associate Professor,
Department of Medicine, Government Medical College, Nagpur

Address for Correspondence -
Dr. Nikhil Nagre
E-mail : mbbs110100@kem.edu

Received on 20th December 2020 Accepted on 23rd December 2020

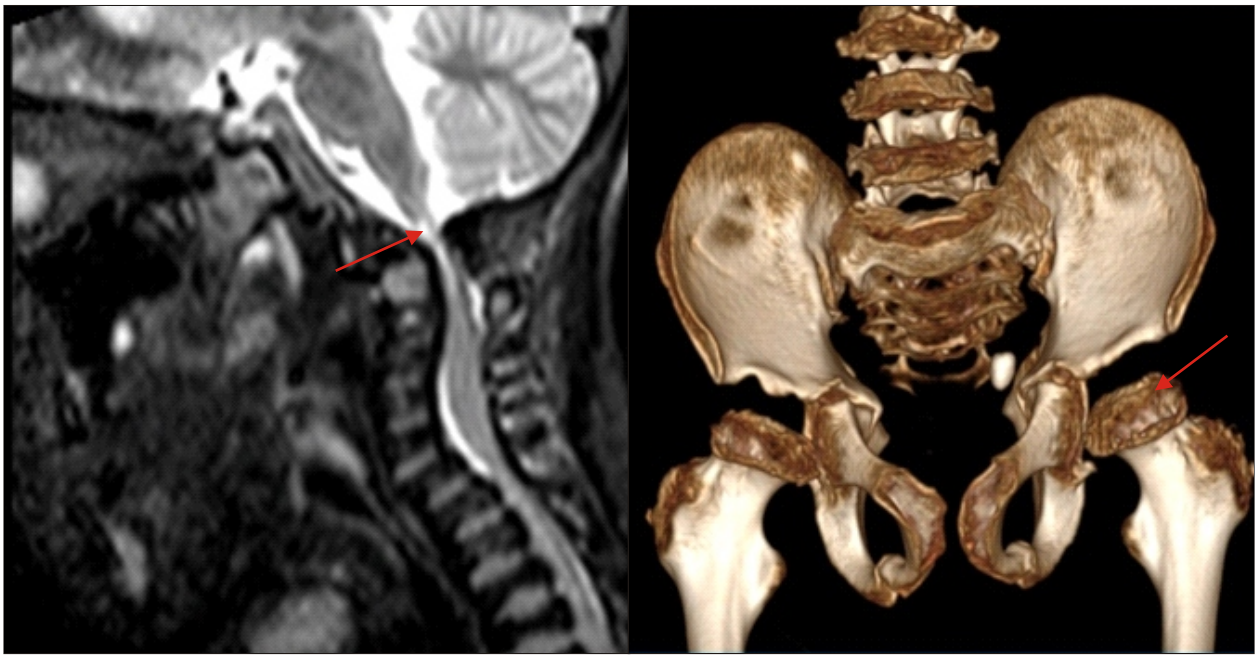
Measurement of glycosaminoglycans in urine was done for differential diagnosis from Morquio Disease [Mucopolysaccharidosis - Type 4] which was within normal range. The diagnosis of SED was made based on dwarfism with a short trunk, characteristic radiographic features and normal urinal levels of GAGs. MRI whole spine screening showed syringomyelia with severe spinal canal stenosis, at C1-2 vertebral level along with myelomalacia and exaggerated lumbar lordosis. Bilateral femoral epiphysis were flattened and dysplastic.



Radiograph of patient s/o epiphyseal dysplasia and flattening

Discussion :

Spondylo-Epiphyseal-Dysplasia-Congenita (SEDC) is a rare autosomal dominant inherited chondrodysplasia. It was first described by Spranger and Wiedemann in 1966[3]. Children with Spondylo-Epiphyseal-Dysplasia-Congenita (SEDC) do present with a wide spectrum of orthopedic abnormalities. Angular deformity of the lower limbs, particularly genu valgum. Lumbar lordosis is an apparent abnormality which in fact mostly related to hip flexion contractures. Coxa vara leads to waddling gait. The latter has been confused by some physicians with myopathy or Mucopolysaccharidosis. Spondylo-Epiphyseal-Dysplasia-Congenita (SEDC) is an autosomal dominant disorder linked to mutation in COL2A1. The disease is characterized by small stature of pre and postnatal onset. Disproportionate short stature with short trunk, flat face, hypertelorism, cleft palate, myopia, vitreoretinal degeneration and retinal detachment at time of growth spurt. Short neck, cervical myopathy (C1-2 instability), barrel-shaped thorax and increased thoracic kyphosis and marked lumbar lordosis with short spine are evident



MRI spine of the patient s/o syrinx with myelomalacia, platyspondyly and epiphyseal dysplasia of femoral heads.

features. The limbs are short, the musculo skeletal phenotype is characterized by hypotonia and waddling gait[4]. Onset is at birth, but severe short stature may not be obvious until 2 to 3 years. In infancy the vertebral bodies are ovoid or pear-shaped but later platyspondyly with irregular endplates develops. Bone age is markedly delayed and the epiphyses are flattened and fragmented. The capital femoral epiphysis is severely affected. The femoral heads might be absent. Delay in ossification of the pubic rami is characteristic. SEDC is a heritable bone dysplasia which mostly results from random mutations sparsely distributed in the 54 exons of the COL2A1 gene[5]. The genetic defect is in the COL2A1 gene located on chromosome 12q13.1-q13.2, and it results in defective procollagen type 2 subunits[6]. Many patients with SEDC, presented with C1-2 instability which might progress to subluxation / dislocation and subsequently lead to cervical myelopathy and quadriparesis with potentially lethal outcome. Spinal cord injury may lead to quadriplegia and sudden death due to respiratory failure. The instability can occur due to odontoid hypoplasia, osodontoideum (OsO) and/or ligamentous laxity. It was noted that C1 inner diameter is smaller than in healthy people, which further compromised the space available for the cord (SAC) at this level in cases of instability[8]. In neonatal and infancy the radiographic manifestations are ovoid or pear-shaped vertebrae. Generalized shortness of the long bones with normal modelling is evident. The pubic and the ischium are hypoplastic. Ossification defects along the epiphyses at hips, knees and no talus or calcaneal ossification. In childhood and later in life, platyspondyly, odontoid hypoplasia and C1-2 instability are characteristic features. The epiphyses are characterized by delay in maturation and irregularity. Restrictive lung disease, laryngeal hypoplasia are additional pathologies[7]. Radiographic findings include the delayed appearance of the epiphysis. Incomplete ossification, coexisting osseous anomalies and angular deformity in SEDC patients may have been an important factors in the high frequency of atlantoaxial dislocation and myelopathy occurrence[7,8]. Atlantoaxial dislocation was initially classified by Greenberg into 2 subcategories reducible and irreducible. Greenberg further devised a treatment strategy based on this system.

For irreducible atlantoaxial dislocation, Greenberg specifically stated that the treatment must be aimed at immediate decompression and achieving stabilization Greenberg work has been considered a landmark publication and is considered by many to be the gold standard for atlantoaxial dislocation treatment[9].

Conclusion :

This case report shows that SEDC patients with CVJ anomalies often have myelopathy, atlantooccipital dissociation due to incomplete ossification, coexisting osseous anomalies and angular deformity. These patients require early aggressive surgical treatment.

References :

1. Spranger JW, Langer LO : Spondyloepiphyseal dysplasia congenita. Radiology 1970; 94: 313-322.
2. Ikegawa S: Spondyloepiphyseal dysplasia congenital.Shouninaika 1998; 30: 294-297
3. Spranger J, Wiedemann H-R. Dysplasia spondyloepiphysaria congenita. Lancet 1966;288:642.
4. Berceanu C, Gheonea AI, Vlaȼdaȼreanu S, et al. Ultrasound and MRI comprehensive approach in prenatal diagnosis of fetalosteochondrodysplasias. Cases series. Med Ultrason 2017;19:66-72.
5. Jung SC, Mathew S, Li QW, et al. Spondyloepiphyseal dysplasia congenita with absent femoral head. J PediatrOrthop B 2004;13:63-9.
6. Tiller GE, Weis AM, Polumbo AP, et al. An RNA-splicing mutation (G +5IVS20) in the type II collagen gene (COL2A1) in a family with spondyloepiphyseal dysplasia congenita. Am J Hum Genet 1995;56:388-95.
7. Miyoshi K, Nakamura K, Haga N, et al. Surgical treatment for atlantoaxial subluxation with myelopathy in spondyloepiphyseal dysplasia congenita. Spine (Phila Pa 1976) 2004;29:E488-91.
8. McKay SD, Al-Omari A, Tomlinson LA, et al. Review of cervical spine anomalies in genetic syndromes. Spine (Phila Pa 1976) 2012;37:E269-77.
9. Greenberg AD. Atlanto-axial dislocations. Brain 1968;91:655-84.